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# Repeat-primed PCR amplification linked to semi-automatic computational analyses showed hexanucleotide expansions of *C9ORF72* gene in a Cohort of Brazilian Amyotrophic Lateral Sclerosis patient

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Genetic mutations are the defined risk factors for Amyotrophic Lateral Sclerosis (ALS) up to date. Despite inherited mutations are linked to Familial forms (FALS) of the disease, specific gene changes may also be encountered in Sporadic forms (SALS). The possibility of genetic mutations may be linked to neuronal degeneration in ALS has stimulated populational studies. Nevertheless, there is a lack of mutational analyses in highly miscegenated populations, as it is Brazil. Mutation in the open reading frame 72 of chromosome 9 (*C9ORF72*) has emerged as one of the most prevalent gene change related to FALS in Europe and North America countries. The mutation is related to a GGGGCC hexanucleotide repeat expansion found in the non-coding region of *C9ORF72* gene was identified in approximately 40% of patients with FALS and 7% of SALS worldwide. The number of repeats is highly variable and may be related to disease evolution. The accurate method to identity mutation in the *C9ORF72* and to count the number of repeats is under continuous development, because nucleotide repeats-linked to neurological disorders are quite common. Classically, they are evaluated by means of southern blot technique followed by autoradiogram quantifications. More recently the repeat-primed polymerase chain reaction amplification, employing a fluorophore-conjugated primer system, linked to semi-automatic computational analyses of fragment length of the hexanucleotide expansion and peaks counting has been developed and has improved the accuracy of analysis. We evaluated in the subjects of our cohort of Brazilian ALS patients the effects of different built-in digital panels, constructed from different sampled subjects, on the morphology of the peaks of fluorescent intensity of every 6 base-pair periodicity. The number of repeat-expansions were determined and submitted to statistical analysis. Pathogenic and non pathogenic *C9ORF72* repeats were found in FALS and SALS of our cohort. Supported by: FAPESP and CNPq, Brazil.

## Biography

Gerson Chadi is Full Professor at the Department of Neurology of the University of Sao Paulo Medical School (USP), Brazil, since 1998. He specialized in Neurodegenerative Diseases and Advanced Research on Regeneration of Central Nervous System (CNS) at Karolinska Institute, Sweden (1991-1994), and also at the Clarke Inst of Psychiatry of Univ of Toronto and at the Montreal Neurological Inst of the McGill Univ (1999). He introduced in Brazil the concepts and research methods in the CNS Regeneration (1994) and was one of the signatories of the Beijing Letter which created the Neurorestauratology Discipline in the field of Neurology. He heads the Translational Neurology Unit, the Translational Neurology Laboratory and the Neuroregeneration Research Center at Dep of Neurology of USP. He coordinates the Neurological Genomic Project and the Cell Therapy Project of his Medical School. He introduced the first Brazilian Systematic Translational Research on Amyotrophic Lateral Sclerosis (ALS, bench to bed, side 2010), being responsible for various clinical and laboratory projects on ALS research. He published more than 100 scientific peer reviewed papers in International Scientific Journals, graduated over than 60 Master, PhD and post doctoral students.

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